Cordlife Carrier Screening Service - Diseases List
康盛人生隱性疾病基因檢測服務- 疾病列表

Related Diseases	相關疾病	Gene
Achalasia-Addisonianism-Alacrima Syndrome	Achalasia-Addisonianism-Alacrima綜合症	AAAS
Harlequin ichthyosis	斑色魚鱗癬	ABCA12
Stargardt Disease, Type 1	1 型斯特格病變	ABCA4
Progressive Familial Intrahepatic Cholestasis, Type 2	2 型進行性遺傳性肝內膽汁淤積症	ABCB11
Progressive Familial Intrahepatic Cholestasis, Type 3	3 型進行性遺傳性肝內膽汁淤積症	ABCB4
Pseudoxanthoma elasticum	彈性纖維偽黃瘤	ABCC6
Familial Hyperinsulinism, ABCC8-Related	遺傳性高胰島素血症(ABCC8 相關)	ABCC8
Adrenoleukodystrophy, X-Linked	X染色體腎上腺腦白質營養不良	ABCD1
Mitochondrial Complex I Deficiency, ACAD9-Related	線粒體複合體 I 缺陷 (ACAD9 相關)	ACAD9
Medium Chain Acyl-CoA Dehydrogenase Deficiency	中鏈酰基輔酶 A 脫氫酶缺乏症	ACADM
Short Chain Acyl-CoA Dehydrogenase Deficiency	短鏈酰基輔酶 A 脫氫酶缺乏症	ACADS
Short/branched chain acyl-CoA dehydrogenase	短鏈/支鏈酰基輔酶 A 脫氫酶	ACADSB
Very Long-Chain Acyl-CoA Dehydrogenase Deficiency	極長鏈酰基輔酶 A 脫氫酶缺乏症	ACADVL
Beta-Ketothiolase Deficiency	β-酮硫解酶缺乏症	ACAT1
Acyl-CoA Oxidase I Deficiency	酰基輔酶 A 氧化酶 I 缺乏症	ACOX1
Combined Malonic and Methylmalonic Aciduria	複合型丙二酸和甲基丙二酸尿症	ACSF3
Severe Combined Immunodeficiency, ADA-Related	嚴重複合型免疫缺陷(ADA 相關)	ADA
Ehlers-Danlos Syndrome, Type VIIC	VIIC 型Ehlers-Danlos 綜合症	ADAMTS2
Bilateral Frontoparietal Polymicrogyria	雙側額葉多小腦回畸形	ADGRG1
Aspartylglucosaminuria	天冬氨酰氨基葡萄糖尿症	AGA
Glycogen Storage Disease, Type III (Cori/Forbes)	3型糖原貯積病 (Cori/Forbes)	AGL
Rhizomelic Chondrodysplasia Punctata, Type 3	3 型肢近端型點狀軟骨發育不良	AGPS
Hyperoxaluria, Primary, Type 1	1 型原發性高草酸尿症	AGXT
Autoimmune polyendocrinopathy syndrome, type I	1型自身免疫性多內分泌病綜合症	AIRE
Sjogren-Larsson Syndrome	Sjogren-Larsson綜合症	ALDH3A2
Pyridoxine-dependent epilepsy	吡哆醇依賴性癲癇	ALDH7A1
Hereditary Fructose Intolerance	遺傳性果糖不耐受症	ALDOB
Congenital Disorder of Glycosylation, Type 1C	1C 型先天性糖基化障礙	ALG6
Alstrom Syndrome	Alstrom綜合症	ALMS1
Hypophosphatasia, ALPL-Related	低磷酸酯酶症(ALPL 相關)	ALPL
Persistent Müllerian duct syndrome type 1	1 型持續性苗勒管綜合症	AMH
Persistent Müllerian duct syndrome type 2	2 型持續性苗勒管綜合症	AMHR2
Glycine Encephalopathy, AMT-Related	甘氨酸腦病 (AMT 相關)	AMT
Mental retardation, enteropathy, deafness, peripheral neuropat	精神發育遲滯、腸病、耳聾、周圍神經病、魚鱗病和角化病 (MED	AP1S1
Familial Nephrogenic Diabetes Insipidus, AQP2-Related	遺傳性腎源性尿崩症(AQP2 相關)	AQP2
Androgen insensitivity syndrome, X-Linked	X染色體雄激素不敏感綜合徵	AR
Argininemia	精氨酸血症	ARG1
Metachromatic Leukodystrophy, ARSA-Related	異染性腦白質營養不良(ARSA 相關)	ARSA
Mucopolysaccharidosis, Type VI (Maroteaux-Lamy)	VI 型粘多醣貯積症（Maroteaux-Lamy）	ARSB
Argininosuccinate Lyase Deficiency	精氨基琥珀酸裂解酶缺乏症	ASL
Asparagine Synthetase Deficiency	天冬酰胺合成酶缺乏症	ASNS
Canavan Disease	卡納文病	ASPA
Citrullinemia, Type 1	1 型瓜氨酸血症	ASS1
Ataxia-Telangiectasia	共濟失調-毛細血管擴張症	ATM
Renal Tubular Acidosis and Deafness, ATP6V1B1-Related	腎小管性酸中毒和耳聾 (ATP6V1B1 相關)	ATP6V1B1
Menkes Syndrome, X-Linked	X染色體門克斯綜合症	ATP7A
Wilson Disease	威爾遜病	ATP7B
Progressive Familial Intrahepatic Cholestasis, Type 1	1 型進行性遺傳性肝內膽汁淤積症	ATP8B1
Alpha-Thalassemia Intellectual Disability Syndrome, X-Linked	X 染色體α-地中海貧血智力障礙綜合症	ATRX
Bardet-Biedl Syndrome 1	Bardet-Biedl 綜合症 1	BBS1
Bardet-Biedl Syndrome 10	Bardet-Biedl 綜合症 10	BBS10
Bardet-Biedl Syndrome 12	Bardet-Biedl 綜合症 12	BBS12
Bardet-Biedl Syndrome 2	Bardet-Biedl 綜合症 2	BBS2
Bardet-Biedl Syndrome 4	Bardet-Biedl 綜合症 4	BBS4
Bardet-Biedl Syndrome 9	Bardet-Biedl 綜合症 9	BBS9
Pseudocholinesterase Deficiency	假膽鹼酯酶缺乏症	BCHE
Maple Syrup Urine Disease, Type 1A	1A 型楓糖漿尿病	BCKDHA
Maple Syrup Urine Disease, Type 1B	1B 型楓糖漿尿病	BCKDHB
GRACILE Syndrome	GRACILE 綜合症	BCS1L
Bloom Syndrome	Bloom綜合症	BLM
Fanconi anemia, Group J	范康尼貧血，J 組	BRIP1

Bartter syndrome, Type 4a	4a 型Bartter 綜合症	BSND
Biotinidase Deficiency	生物素酶缺乏症	BTD
Isolated growth hormone deficiency, Type III, X-linked	X 染色體III 型孤立性生長激素缺乏症	BTK
Desbuquois dysplasia 1	Desbuquois 發育不良 1	CANT1
Limb-Girdle Muscular Dystrophy, Type 2A	2A 型肢帶型肌營養不良症	CAPN3
Catecholaminergic polymorphic ventricular tachycardia	兒茶酚胺能多形性室性心動過速	CASQ2
Homocystinuria, CBS-Related	CBS 相關同型半胱氨酸尿症	CBS
Mental retardation, autosomal recessive 3	智力障礙，常染色體隱性遺傳 3	CC2D1A
Usher Syndrome, Type 1D	1D 型Usher 綜合症	CDH23
Leber Congenital Amaurosis, Type CEP290	Leber 先天性黑朦症 (CEP290 型)	CEP290
Retinitis Pigmentosa 26	色素性視網膜炎 26	CERKL
Cystic Fibrosis	囊性纖維化	CFTR
Choroideremia, X-Linked	X染色體無脈絡膜	CHM
Congenital Myasthenic Syndrome, CHRNE-Related	先天性肌無力綜合徵(CHRNE 相關)	CHRNE
Escobar Syndrome	埃斯科巴綜合症	CHRNG
Bare Lymphocyte Syndrome, CIITA-Related	裸淋巴細胞綜合症 (CIITA 相關)	CIITA
Ceroid Lipofuscinosis, Neuronal, 3	類固醇脂褐質沉著症，神經元3	CLN3
Ceroid Lipofuscinosis, Neuronal, 5	類固醇脂褐質沉著症，神經元5	CLN5
Ceroid Lipofuscinosis, Neuronal, 6	類固醇脂褐質沉著症，神經元6	CLN6
Ceroid Lipofuscinosis, Neuronal, 8 (a.ka. Northern Epilepsy)	類固醇脂褐質沉著症，神經元8（又名北方癲癇症）	CLN8
Usher Syndrome, Type 3	3 型Usher綜合症	CLRN1
Achromatopsia, CNGA3-Related	全色盲(CNGA3 相關)	CNGA3
Achromatopsia, CNGB3-Related	全色盲(CNGB3 相關)	CNGB3
Fibrochondrogenesis type 2	2 型纖維軟骨形成	COL11A2
Alport Syndrome, COL4A3-Related	Alport 綜合症(COL4A3 相關)	COL4A3
Alport Syndrome, COL4A4-Related	Alport 綜合症 (COL4A4 相關)	COL4A4
Alport Syndrome, X-Linked	X 染色體Alport 綜合症	COL4A5
Dystrophic Epidermolysis Bullosa, COL7A1-Related	營養不良性大皰性表皮鬆解症(COL7A1 相關)	COL7A1
Carbamoyl Phosphate Synthetase I Deficiency	氨基甲酰磷酸合成酶 I 缺乏症	CPS1
Carnitine Palmitoyltransferase IA Deficiency	肉鹼棕櫚酰轉移酶 IA 缺乏症	CPT1A
Carnitine Palmitoyltransferase II Deficiency	肉鹼棕櫚酰轉移酶 II 缺乏症	CPT2
Leber congenital amaurosis 8	Leber 先天性黑矇症 8	CRB1
Cystinosis	胱氨酸增多症	CTNS
Papillon-Lefevre Syndrome	Papillon-Lefevre 綜合症	CTSC
Ceroid Lipofuscinosis, Neuronal, 10 (CLN10 Disease)	類固醇脂褐質沉著症，神經元10（CLN10 病變）	CTSD
Pycnodysostosis	緻密性成骨不全症	CTSK
Chronic Granulomatous Disease, CYBA-Related	慢性肉芽腫病(CYBA 相關)	CYBA
Chronic Granulomatous Disease, X-Linked	X染色體慢性肉芽腫病	CYBB
Congenital Adrenal Hyperplasia, 11-beta-hydroxylase-deficien	先天性腎上腺增生，11-β-羥化酶缺陷	CYP11B1
Corticosterone Methyloxidase Deficiency	皮質酮甲基氧化酶缺乏症	CYP11B2
Congenital Adrenal Hyperplasia, 17-Alpha-Hydroxylase Deficie	先天性腎上腺增生症，17-α-羥化酶缺乏症	CYP17A1
Aromatase Deficiency	芳香酶缺乏症	CYP19A1
Primary Congenital Glaucoma	原發性先天性青光眼	CYP1B1
Congenital Adrenal Hyperplasia, 21-hydroxylase-deficient	先天性腎上腺增生症，21-羥化酶缺陷型	CYP21A2
Cerebrotendinous Xanthomatosis	腦腱黃瘤病	CYP27A1
Vitamin D-dependent rickets type 1A	1A 型維他命D 依賴性佝僂病	CYP27B1
Maple Syrup Urine Disease, Type 2	2 型楓糖漿尿病	DBT
Severe Combined Immunodeficiency, Type Athabaskan	Athabaskan 型嚴重聯合免疫缺陷	DCLRE1C
Xeroderma Pigmentosum Group E	著色性乾皮病E組	DDB2
Smith-Lemli-Opitz Syndrome	Smith-Lemli-Opitz 綜合症	DHCR7
Retinitis Pigmentosa 59	色素性視網膜炎 59	DHDDS
Dyskeratosis congenita, X-Linked	X染色體先天性角化不良	DKC1
Dihydrolipoamide Dehydrogenase Deficiency	二氫硫辛酰胺脫氫酶缺乏症	DLD
Duchenne/Becker Muscular Dystrophy	Duchenne/Becker 肌營養不良症	DMD
Ciliary Dyskinesia, Primary 3	纖毛運動障礙，先天3型	DNAH5
Ciliary Dyskinesia, Primary 1	纖毛運動障礙，先天1型	DNAI1
Ciliary Dyskinesia, Primary 9	纖毛運動障礙，先天9型	DNAI2
Ciliary Dyskinesia, Primary, 16	纖毛運動障礙，先天16型	DNAL1
Congenital Myasthenic Syndrome, DOK7-Related	先天性肌無力綜合症 (DOK7 相關)	DOK7
Dihydropyrimidine Dehydrogenase Deficiency	二氫嘧啶脫氫酶缺乏症	DPYD
Limb-Girdle Muscular Dystrophy, Type 2B	2B 型肢帶型肌營養不良症	DYSF
Hypohidrotic Ectodermal Dysplasia, X-Linked	X 染色體少汗性外胚層發育不良	EDA
Hypohidrotic Ectodermal Dysplasia	少汗性外胚層發育不良	EDAR
Wolcott-Rallison Syndrome	Wolcott-Rallison 綜合症	EIF2AK3
Leukoencephalopathy with Vanishing White Matter	白質消失的白質腦病	EIF2B5
Dysautonomia, familial (IKBKAP or ELP1)	遺傳性自主神經障礙（IKBKAP 或 ELP1）	ELP1

Emery-Dreifuss Muscular Dystrophy 1, X-Linked	X 染色體Emery-Dreifuss 肌肉萎縮症1	EMD
Xeroderma Pigmentosum Group D	色素性乾皮病D組	ERCC2
Xeroderma Pigmentosum Group B	著色性乾皮病B組	ERCC3
Xeroderma Pigmentosum Group F	著色性乾皮病F組	ERCC4
Xeroderma pigmentosum Group G	著色性乾皮病G組	ERCC5
Cockayne syndrome, type B	B 型Cockayne 綜合症	ERCC6
Cockayne syndrome, type A	A 型Cockayne 綜合症	ERCC8
Roberts Syndrome	Roberts綜合症	ESCO2
Glutaric Acidemia, Type 2A	2A 型戊二酸血症	ETFA
Glutaric Acidemia, Type 2B	2B 型戊二酸血症	ETFB
Glutaric Acidemia, Type 2C	2C 型戊二酸血症	ETFDH
Ethylmalonic Encephalopathy	乙基丙二酸腦病	ETHE1
Ellis-van Creveld Syndrome, EVC-Related	Ellis-van Creveld 綜合症 (EVC 相關)	EVC
Ellis-van Creveld Syndrome, EVC2-related	Ellis-van Creveld 綜合症 (EVC2 相關)	EVC2
Pontocerebellar Hypoplasia, Type 1B	1B 型腦橋小腦發育不全	EXOSC3
Retinitis Pigmentosa 25	色素性視網膜炎 25	EYS
Factor XI deficiency	因子 XI 缺乏症	F11
Prothrombin deficiency	凝血酶原缺乏症	F2
Hemophilia A	甲型血友病	F8
Hemophilia B	乙型血友病	F9
Tyrosinemia, Type I	1型酪氨酸血症	FAH
Retinitis Pigmentosa 28	色素性視網膜炎 28	FAM161A
Fanconi Anemia, Group A	范康尼貧血，A 組	FANCA
Fanconi Anemia, Group C	范康尼貧血，C 組	FANCC
Fanconi Anemia, Group G	范康尼貧血，G 組	FANCG
Fumarase Deficiency	富馬酸酶缺乏症	FH
Limb-Girdle Muscular Dystrophy, Type 2I	2I 型肢帶型肌肉萎縮症	FKRP
Walker-Warburg Syndrome, FKTN-Related	Walker-Warburg 綜合症 (FKTN 相關)	FKTN
Fragile X syndrome	脆性 X 綜合症	FMR1
Glycogen Storage Disease, Type IA	IA 型糖原貯積病	G6PC
Glucose-6-Phosphate Dehydrogenase Deficiency	6-磷酸葡萄糖脫氫酶缺乏症	G6PD
Glycogen Storage Disease, Type II (Pompe Disease)	2型糖原貯積病（龐貝病）	GAA
Krabbe Disease	克拉伯病	GALC
Galactose epimerase deficiency	半乳糖差向異構酶缺乏症	GALE
Galactokinase Deficiency (Galactosemia, Type II)	半乳糖激酶缺乏症（半乳糖血症，II 型）	GALK1
Mucopolysaccharidosis, Type IVA	黏多醣貯積症，IVA型	GALNS
Hyperphosphatemic familial tumoral calcinosis	高磷血症遺傳性腫瘤性鈣質沉著症	GALNT3
Galactosemia	半乳糖血症	GALT
Guanidinoacetate Methyltransferase Deficiency	胍基乙酸甲基轉移酶缺乏症	GAMT
Gaucher Disease	高雪氏症	GBA
Glycogen Storage Disease, Type IV	IV 型糖原貯積病	GBE1
Glutaric Acidemia, Type 1	1 型戊二酸血症	GCDH
Dopa-responsive dystonia	多巴反應性肌張力障礙	GCH1
Grebe syndrome	Grebe綜合症	GDF5
Combined Oxidative Phosphorylation Deficiency 1	聯合氧化磷酸化缺陷 1	GFM1
Isolated growth hormone deficiency, Type IA/II	IA/II 型孤立性生長激素缺乏症	GH1
Isolated growth hormone deficiency, Type IB	IB 型孤立性生長激素缺乏症	GHRHR
Charcot-Marie-Tooth Disease with Deafness, X-Linked	X 染色體伴有耳聾的 Charcot-Marie-Tooth 病變	GJB1
Non-Syndromic Hearing Loss (a.k.a. Connexin 26)	非綜合徵性聽力損失（又名連接蛋白 26）	GJB2
Erythrokeratodermia variabilis et progressiva	變異性和進行性紅角化病	GJB3
Non-Syndromic Hearing Loss (a.k.a. Connexin 30)	非綜合徵性聽力損失（又名連接蛋白 30）	GJB6
Fabry Disease	Fabry病變	GLA
Mucopolysaccharidosis, Type IVB / GM1 Gangliosidosis	粘多醣貯積症，IVB 型/GM1 型神經節苷脂貯積症	GLB1
Glycine Encephalopathy, GLDC-Related	甘氨酸腦病 (GLDC 相關)	GLDC
Lethal Congenital Contracture Syndrome 1	致死性先天性攣縮綜合症 1	GLE1
Inclusion Body Myopathy 2	包涵體肌病 2	GNE
Mucolipidosis II/IIIA	粘脂沉積症 II/IIIA	GNPTAB
Mucolipidosis III gamma	黏脂沉積症 III γ	GNPTG
Mucopolysaccharidosis, Type IIID (Sanfilippo D)	粘多醣貯積症，IIID 型 (Sanfilippo D)	GNS
Geroderma osteodysplastica	骨發育不良白皮病	GORAB
Bernard-Soulier Syndrome, Type A2	A2 型Bernard-Soulier 綜合症	GP1BA
Bernard-Soulier Syndrome, Type B	B 型Bernard-Soulier 綜合症	GP1BB
Bernard-Soulier Syndrome, Type C	C 型Bernard-Soulier 綜合症	GP9
Primary Hyperoxaluria, Type 2	2 型原發性高草酸尿症	GRHPR
Leber congenital amaurosis 1	Leber先天性黑朦症1	GUCY2D
Mucopolysaccharidosis, Type VII	VII 型粘多醣貯積症	GUSB

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency	長鏈 3-羥酰基輔酶 A 脫氫酶缺乏症	HADHA
Trifunctional protein deficiency	三功能蛋白缺乏症	HADHB
Congenital Neutropenia, HAX1-Related	先天性中性粒細胞減少症(HAX1 相關)	HAX1
Alpha-Thalassemia	α-地中海貧血	HBA1
Alpha-Thalassemia	α-地中海貧血	HBA2
Beta-Hemoglobinopathies	β-血紅蛋白病	HBB
Tay-Sachs Disease	Tay-Sachs病變	HEXA
Sandhoff Disease	Sandhoff病變	HEXB
Hemochromatosis, Type 1	1 型血色素沉著症	HFE
Hemochromatosis, Type 2A	2A 型血色素沉著症	HFE2
Alkaptonuria	黑尿症	HGD
Mucopolysaccharidosis, Type IIIC (Sanfilippo C)	IIIC 型黏多醣貯積症 (Sanfilippo C)	HGSNAT
Holocarboxylase Synthetase Deficiency	全羧化酶合成酶缺乏症	HLCS
3-Hydroxy-3-Methylglutaryl-Coenzyme A Lyase Deficiency	3-羥基-3-甲基戊二酰輔酶 A 裂解酶缺乏症	HMGCL
Heme Oxygenase-1 Deficiency	血紅素加氧酶 1 缺乏症	HMOX1
Primary Hyperoxaluria, Type 3	3 型原發性高草酸尿症	HOGA1
Tyrosinemia, Type III	III 型酪氨酸血症	HPD
Hermansky-Pudlak Syndrome 1	Hermansky-Pudlak 綜合症 1	HPS1
Hermansky-Pudlak Syndrome 3	Hermansky-Pudlak 綜合症 3	HPS3
Hermansky-Pudlak syndrome 4	Hermansky-Pudlak 綜合症 4	HPS4
17-beta hydroxysteroid dehydrogenase 3 deficiency	17-β 羥基類固醇脫氫酶 3 缺乏症	HSD17B3
D-Bifunctional Protein Deficiency	D-雙功能蛋白缺乏症	HSD17B4
3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency	3-β-羥基類固醇脫氫酶 II 型缺乏症	HSD3B2
Hydrolethalus Syndrome	Hydrolethalus綜合症	HYLS1
Mucopolysaccharidosis, Type II (Hunter Syndrome)	II 型黏多醣貯積症（Hunter綜合症）	IDS
Mucopolysaccharidosis, Type I (Hurler Syndrome)	I 型粘多醣貯積症（Hurler 綜合症）	IDUA
Severe Combined Immunodeficiency, X-Linked	X 染色體嚴重複合型免疫缺陷	IL2RG
Glanzmann thrombasthenia	格蘭茨曼血小板無力症	ITGB3
Isovaleric Acidemia	異戊酸血症	IVD
Congenital Hyperinsulinism, KCNJ11-Related	先天性高胰島素血症 (KCNJ11 相關)	KCNJ11
LAMA2-related Muscular Dystrophy	肌肉萎縮症 (LAMA2相關)	LAMA2
Herlitz Junctional Epidermolysis Bullosa, LAMA3-Related	Herlitz 交界性大皰性表皮鬆解症 (LAMA3 相關)	LAMA3
Herlitz Junctional Epidermolysis Bullosa, LAMB3-Related	Herlitz 交界性大皰性表皮鬆解症 (LAMB3 相關)	LAMB3
Herlitz Junctional Epidermolysis Bullosa, LAMC2-Related	Herlitz 交界性大皰性表皮鬆解症 (LAMC2 相關)	LAMC2
Leber Congenital Amaurosis, Type LCA5	LCA5型Leber先天性黑朦症	LCA5
Familial Hypercholesterolemia, LDLR-Related	遺傳性高膽固醇血症 (LDLR 相關)	LDLR
Familial Hypercholesterolemia, LDLRAP1-Related	遺傳性高膽固醇血症 (LDLRAP1 相關)	LDLRAP1
Leydig cell hypoplasia	間質細胞發育不全	LHCGR
Stuve-Wiedemann Syndrome	Stuve-Wiedemann 綜合症	LIFR
Lysosomal Acid Lipase Deficiency	溶酶體酸性脂肪酶缺乏症	LIPA
Woolly Hair/Hypotrichosis Syndrome	毛髮/少毛症綜合症	LIPH
Deafness, Autosomal Recessive 77	耳聾 (常染色體隱性遺傳 77)	LOXHD1
Lipoprotein Lipase Deficiency	脂蛋白脂肪酶缺乏症	LPL
Leigh Syndrome, French-Canadian Type	Leigh 綜合症，法裔加拿大人型	LRPPRC
Chediak-Higashi syndrome	Chediak-Higashi綜合症	LYST
Alpha-Mannosidosis	α-甘露糖苷貯積症	MAN2B1
Hypermethioninemia	高蛋氨酸血症	MAT1A
3-Methylcrotonyl-CoA Carboxylase 1 Deficiency	3-甲基巴豆酰輔酶 A 羧化酶 1 缺乏症	MCCC1
3-Methylcrotonyl-CoA Carboxylase 2 Deficiency	3-甲基巴豆酰輔酶 A 羧化酶 2 缺乏症	MCCC2
Mucolipidosis, Type IV	IV 型粘脂沉積症	MCOLN1
RETT Syndrome	雷特綜合症	MECP2
Microcephaly, postnatal progressive, with seizures and brain at	小頭畸形，產後進行性發作，伴有癲癇發作和腦萎縮	MED17
Familial Mediterranean Fever	遺傳性地中海熱	MEFV
Spondylothoracic Dysostosis, MESP2-Related	脊椎發育不良 (MESP2 相關)	MESP2
Ceroid Lipofuscinosis, Neuronal, 7	類固醇脂褐質沉著症，神經元7	MFSD8
Bardet-Biedl Syndrome 6	Bardet-Biedl 綜合症 6	MKKS
Meckel-Gruber Syndrome, Type 1	1 型Meckel-Gruber 綜合症	MKS1
Megalencephalic Leukoencephalopathy with Subcortical Cysts	伴皮質下囊腫的巨腦性白質腦病	MLC1
Malonyl-CoA decarboxylase deficiency	丙二酰輔酶A脫羧酶缺乏症	MLYCD
Methylmalonic Aciduria, MMAA-Related	甲基丙二酸尿症 (MMAA 相關)	MMAA
Methylmalonic Aciduria, MMAB-Related	甲基丙二酸尿症 (MMAB 相關)	MMAB
Methylmalonic Aciduria and Homocystinuria, Type cblC	甲基丙二酸尿症和高胱氨酸尿症 (cblC 型)	MMACHC
Methylmalonic Aciduria and Homocystinuria, Type cblD	甲基丙二酸尿症和高胱氨酸尿症 (cblD 型)	MMADHC
Molybdenum cofactor deficiency	鉬輔助因子缺乏症	MOCS1
Congenital Disorder of Glycosylation, Type 1B	1B 型先天性糖基化障礙	MPI
Congenital Amegakaryocytic Thrombocytopenia	先天性無巨核細胞性血小板減少症	MPL

Hepatocerebral Mitochondrial DNA Depletion Syndrome, MPV	肝腦線粒體 DNA 耗竭綜合症 (MPV17 相關)	MPV17
Ataxia-telangiectasia-like disorder 1	共濟失調毛細血管擴張樣疾病1	MRE11
Homocystinuria due to Deficiency of MTHFR	MTHFR 缺乏引起的同型半胱氨酸尿症	MTHFR
Myotubular Myopathy, X-Linked	X染色體肌管性肌病	MTM1
Homocystinuria, Type cblE	cblE 型同型半胱氨酸尿症	MTRR
Abetalipoproteinemia	無β脂蛋白血症	MTTP
Methylmalonic Aciduria, Type mut(0)	甲基丙二酸尿症，類型 mut(0)	MUT
Deafness, autosomal recessive, 3	耳聾，常染色體隱性遺傳，3	MYO15A
Usher Syndrome, Type 1B	1B 型Usher綜合症	MYO7A
Mucopolysaccharidosis, Type IIIB (Sanfilippo B)	IIIB 型粘多醣貯積症 (Sanfilippo B)	NAGLU
N-acetylglutamate Synthase Deficiency	N-乙酰谷氨酸合酶缺乏症	NAGS
Nijmegen Breakage Syndrome	Nijmegen斷裂綜合症	NBN
Charcot-Marie-Tooth Disease type 4D	4D 型Charcot-Marie-Tooth 病	NDRG1
Mitochondrial Complex I Deficiency, NDUFAF5-Related	線粒體複合體 I 缺陷 (NDUFAF5 相關)	NDUFAF5
Mitochondrial complex I deficiency	線粒體複合物 I 缺陷	NDUFS4
Mitochondrial Complex I Deficiency, NDUFS6-Related	線粒體複合體 I 缺陷 (NDUFS6 相關)	NDUFS6
Nemaline Myopathy, NEB-Related	Nemaline 肌病 (NEB 相關)	NEB
Sialidosis	唾液酸增多症	NEU1
Hydatidiform Mole, Recurrent	葡萄胎，復發型	NLRP7
Niemann-Pick Disease, Type C1/D	C1/D 型Niemann-Pick病變	NPC1
Niemann-Pick Disease, Type C2	C2 型Niemann-Pick病變	NPC2
Juvenile Nephronophthisis	幼年腎病	NPHP1
Congenital Finnish Nephrosis	先天性芬蘭腎病	NPHS1
Steroid-Resistant Nephrotic Syndrome	類固醇抵抗性腎病綜合症	NPHS2
Congenital Adrenal Hypoplasia, X-linked	X染色體先天性腎上腺發育不全	NR0B1
Enhanced S-Cone Syndrome	增強型 S 錐綜合症	NR2E3
Congenital Insensitivity to Pain with Anhidrosis (CIPA)	先天性疼痛無汗症 (CIPA)	NTRK1
Ornithine Aminotransferase Deficiency	鳥氨酸氨基轉移酶缺乏症	OAT
Lowe syndrome, X-Linked	X染色體Lowe綜合症	OCRL
Costeff Syndrome (3-Methylglutaconic Aciduria, Type 3)	Costeff 綜合症（3-甲基戊烯酸尿症，3 型）	OPA3
Ornithine Transcarbamylase Deficiency	鳥氨酸轉氨甲酰酶缺乏症	OTC
Phenylketonuria	苯丙酮尿症	PAH
Pantothenate Kinase-Associated Neurodegeneration	泛酸激酶相關的神經變性	PANK2
Pyruvate Carboxylase Deficiency	丙酮酸羧化酶缺乏症	PC
Propionic Acidemia, PCCA-Related	丙酸血症 (PCCA 相關)	PCCA
Propionic Acidemia, PCCB-Related	丙酸血症 (PCCB 相關)	PCCB
Usher Syndrome, Type 1F	1F 型亞瑟綜合症	PCDH15
Pyruvate Dehydrogenase Deficiency, X-Linked	X 染色體丙酮酸脫氫酶缺乏症	PDHA1
Pyruvate Dehydrogenase Deficiency, PDHB-Related	丙酮酸脫氫酶缺乏症 (PDHB 相關)	PDHB
Prolidase deficiency	脯氨酸酶缺乏症	PEPD
Cytochrome-c oxidase deficiency	細胞色素c氧化酶缺乏症	PET100
Peroxisome Biogenesis Disorder 1A (Zellweger)	過氧化物酶體生物發生障礙 1A (Zellweger)	PEX1
Peroxisome Biogenesis Disorder 6A (Zellweger)	過氧化物酶體生物發生障礙 6A (Zellweger)	PEX10
Peroxisome Biogenesis Disorder 3A (Zellweger)	過氧化物酶體生物發生障礙 3A (Zellweger)	PEX12
Peroxisome Biogenesis Disorder 5A (Zellweger)	過氧化物酶體生物發生障礙 5A (Zellweger)	PEX2
Peroxisome Biogenesis Disorder 4A (Zellweger)	過氧化物酶體生物發生障礙 4A (Zellweger)	PEX6
Rhizomelic Chondrodysplasia Punctata, Type 1	1 型肢近端型點狀軟骨發育不良	PEX7
Glycogen Storage Disease, Type VII	VII 型糖原貯積病	PFKM
Phosphoglycerate Dehydrogenase Deficiency	磷酸甘油酸脫氫酶缺乏症	PHGDH
Multiple congenital anomalies-hypotonia-seizures syndrome 1	多發性先天性異常-肌張力減退-癲癇綜合症 1	PIGN
Polycystic Kidney Disease, Autosomal Recessive	常染色體隱性遺傳多囊腎病	PKHD1
Infantile neuroaxonal dystrophy 1	嬰兒神經軸索營養不良 1	PLA2G6
Congenital Disorder of Glycosylation, Type 1A, PMM2-Related	1A 型先天性糖基化障礙 (PMM2 相關)	PMM2
Pyridoxal 5'-phosphate-dependent epilepsy	5'-磷酸吡哆醛依賴性癲癇	PNPO
POLG-Related Disorders	POLG 相關疾病	POLG
Xeroderma pigmentosum Variant	著色性乾皮病變	POLH
Muscle-Eye-Brain Disease, POMGNT1-Related	肌眼腦疾病 (POMGNT1 相關)	POMGNT1
Cytochrome P450 oxidoreductase deficiency	細胞色素 P450 氧化還原酶缺乏症	POR
Ceroid Lipofuscinosis, Neuronal, 1	類固醇脂褐質沉著症，神經元，1	PPT1
Myasthenic syndrome, congenital, 22	肌無力綜合症，先天性22	PREPL
Combined Pituitary Hormone Deficiency 2	聯合垂體激素缺乏症 2	PROP1
Arts syndrome, X-Linked	X染色體Arts綜合症	PRPS1
Metachromatic Leukodystrophy, PSAP-Related	異染性腦白質營養不良 (PSAP 相關)	PSAP
6-Pyruvoyl-Tetrahydropterin Synthase (PTPS) Deficiency	6-丙酮酰-四氫蝶呤合酶 (PTPS) 缺乏症	PTS
Mitochondrial Myopathy and Sideroblastic Anemia (MLASA1)	線粒體肌病和鐵粒幼細胞性貧血 (MLASA1)	PUS1
Glycogen Storage Disease, Type V (McArdle Disease)	V 型糖原貯積病（McArdle病變）	PYGM

Carpenter Syndrome	Carpenter綜合症	RAB23
Omenn Syndrome, RAG1-Related	Omenn 綜合症 (RAG1 相關)	RAG1
Omenn Syndrome, RAG2-Related	Omenn 綜合症 (RAG2 相關)	RAG2
Congenital Myasthenic Syndrome, RAPSN-Related	先天性肌無力綜合症 (RAPSN 相關)	RAPSN
Pontocerebellar Hypoplasia, Type 1 and 6, RARS2-Related	1 型及6 型腦橋小腦發育不全 (RARS2 相關)	RARS2
Leber Congenital Amaurosis, Type RDH12	RDH12 型Leber 先天性黑朦症	RDH12
Retinal Dystrophies, RLBP1-Associated	視網膜營養不良 (RLBP1 相關)	RLBP1
Cartilage-Hair Hypoplasia	軟骨毛髮發育不全	RMRP
Aicardi-Goutieres syndrome, RNASEH2C-related	Aicardi-Goutieres 綜合症 (RNASEH2C 相關)	RNASEH2C
Leber Congenital Amaurosis 2	Leber 先天性黑朦症2	RPE65
Ciliopathies, RPGRIP1L-Related	纖毛病變 (RPGRIP1L 相關)	RPGRIP1L
Juvenile Retinoschisis, X-Linked	X 染色體青少年視網膜劈裂症	RS1
Dyskeratosis Congenita, RTEL1-Related	先天性角化不良 (RTEL1 相關)	RTEL1
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay	Charlevoix-Saguenay 的常染色體隱性痙攣性共濟失調	SACS
MIRAGE syndrome	MIRAGE綜合症	SAMD9
Aicardi-Goutires Syndrome	Aicardi-Goutires綜合症	SAMHD1
Shwachman-Diamond syndrome	Shwachman-Diamond綜合症	SBDS
Pontocerebellar Hypoplasia, Type 2D	2D 型橋腦小腦發育不全	SEPSECS
Alpha-1-Antitrypsin Deficiency	α-1-抗胰蛋白酶缺乏症	SERPINA1
Limb-Girdle Muscular Dystrophy, Type 2D	2D 型肢帶型肌肉萎縮症	SGCA
Limb-Girdle Muscular Dystrophy, Type 2E	2E 型肢帶型肌肉萎縮症	SGCB
Limb-Girdle Muscular Dystrophy, Type 2F	2F 型肢帶型肌肉萎縮症	SGCD
Limb-Girdle Muscular Dystrophy, Type 2C	2C 型肢帶型肌肉萎縮症	SGCG
Mucopolysaccharidosis, Type IIIA (Sanfilippo A)	IIIA 型 粘多醣貯積症 (Sanfilippo A)	SGSH
Gitelman Syndrome	Gitelman綜合症	SLC12A3
Agenesis of the Corpus Callosum with Peripheral Neuropathy (	胼胝體發育不全伴周圍神經病變（Andermann綜合症）	SLC12A6
Salla Disease	Salla 病變	SLC17A5
Megaloblastic Anemia Syndrome	巨幼紅細胞性貧血綜合症	SLC19A2
Carnitine Deficiency	肉鹼缺乏症	SLC22A5
Citrullinemia, Type II	II 型瓜氨酸血症	SLC25A13
Hyperornithinemia-Hyperammonemia-Homocitrullinuria (HHH	高鳥氨酸血症-高氨血症-高瓜氨酸尿症 (HHH) 綜合症	SLC25A15
Carnitine-acylcarnitine translocase deficiency	肉鹼-酰基肉鹼轉位酶缺乏症	SLC25A20
Achondrogenesis, Type 1B	1B 型軟骨發育不全	SLC26A2
Congenital Chloride Diarrhea	先天性氯化物腹瀉	SLC26A3
Pendred Syndrome	Pendred綜合症	SLC26A4
Autism Spectrum, Epilepsy and Arthrogryposis	自閉症、癲癇和關節攣縮症	SLC35A3
Glycogen Storage Disease, Type IB	IB 型糖原貯積病	SLC37A4
Acrodermatitis Enteropathica	腸病性肢端皮炎	SLC39A4
Cystinuria, Type A	A 型胱氨酸尿症	SLC3A1
Oculocutaneous albinism, Type 4	4 型眼皮膚白化病	SLC45A2
Corneal Dystrophy and Perceptive Deafness	角膜營養不良和知覺性耳聾	SLC4A11
Creatine Transporter Defect (Cerebral Creatine Deficiency Synd	肌酸轉運蛋白缺陷（X 染色體腦肌酸缺乏綜合徵 1）	SLC6A8
Lysinuric Protein Intolerance	賴氨酸蛋白不耐受	SLC7A7
Cystinuria, Type B	B 型胱氨酸尿症	SLC7A9
Schimke Immunoosseous Dysplasia	Schimke 免疫骨發育不良	SMARCAL1
Spinal Muscular Atrophy	脊髓性肌萎縮症	SMN1
Niemann-Pick Disease, Types A/B	A/B 型Niemann-Pick病	SMPD1
5-alpha reductase deficiency	5-α還原酶缺乏症	SRD5A2
GM3 synthase deficiency	GM3合酶缺乏症	ST3GAL5
Lipoid Congenital Adrenal Hyperplasia	類脂性先天性腎上腺增生症	STAR
Deafness, autosomal recessive 16	常染色體隱性遺傳 16耳聾	STRC
Mitochondrial DNA depletion syndrome 5 (encephalomyopath	線粒體 DNA 耗竭綜合症 5（腦肌病伴或不伴甲基丙二酸尿症）	SUCLA2
Multiple Sulfatase Deficiency	多種硫酸酯酶缺乏症	SUMF1
Leigh Syndrome	Leigh綜合症	SURF1
Tyrosinemia, Type II	酪氨酸血症，II 型	TAT
Osteopetrosis, Infantile Malignant, TCIRG1-Related	骨硬化症及嬰兒惡性腫瘤 (TCIRG1 相關)	TCIRG1
Hereditary Spastic Paraparesis, Type 49	49 型遺傳性痙攣性截癱	TECPR2
Hemochromatosis, Type 3, TFR2-Related	3 型血色素沉著症 (TFR2 相關)	TFR2
Lamellar Ichthyosis, Type 1	1 型板層狀魚鱗病變	TGM1
Segawa Syndrome, TH-Related	Segawa 綜合症 (TH 相關)	TH
Deafness, autosomal dominant 36, autosomal recessive 7	常染色體顯性遺傳 36，常染色體隱性遺傳 7耳聾	TMC1
Joubert Syndrome 2 / Meckel Syndrome 2	Joubert 綜合症 2 / Meckel 綜合症 2	TMEM216
Congenital hypothyroidism	先天性甲狀腺機能減退症	TPO
Ceroid Lipofuscinosis, Neuronal, 2	類固醇脂褐質沉著症，神經元2	TPP1
Aicardi-Goutieres syndrome, TREX1-related	Aicardi-Goutieres 綜合症 (TREX1 相關)	TREX1
Bardet-Biedl syndrome 11	Bardet-Biedl 綜合症11	TRIM32

Mulibrey nanism syndrome	Mulibrey nanism綜合症	TRIM37
Acute Infantile Liver Failure, TRMU-Related	急性嬰兒肝功能衰竭 (TRMU 相關)	TRMU
Pontocerebellar hypoplasia	腦橋小腦發育不全	TSEN54
Combined Oxidative Phosphorylation Deficiency 3	複合型氧化磷酸化缺陷 3	TSFM
Congenital hypothyroidism	先天性甲狀腺機能減退症	TSHB
Hypothyroidism, congenital, nongoitrous, 1	甲狀腺功能減退症，先天性，非甲狀腺腫，1	TSHR
Tricho-Hepato-Enteric Syndrome	Tricho-Hepato-Enteric 綜合症	TTC37
Familial dilated cardiomyopathy	遺傳性擴張型心肌病變	TTN
Ataxia with Vitamin E Deficiency	維他命 E 缺乏的共濟失調	TTPA
Myoneurogastrointestinal Encephalopathy (MNGIE)	肌神經胃腸腦病變 (MNGIE)	TYMP
Oculocutaneous Albinism, Type 1	1 型眼皮膚白化病	TYR
Oculocutaneous albinism, Type 3	3 型眼皮膚白化病	TYRP1
Crigler-Najjar Syndrome	Crigler-Najjar綜合症	UGT1A1
Beta-ureidopropionase deficiency	β-脲基丙酸酶缺乏症	UPB1
Usher Syndrome, Type 1C	1C 型Usher綜合症	USH1C
Usher Syndrome, Type 2A	2A 型Usher綜合症	USH2A
Choreo-acanthocytosis	舞蹈性棘紅細胞增多症	VPS13A
Cohen Syndrome	Cohen 綜合症	VPS13B
Congenital Neutropenia, VPS45-Related	VPS45 相關先天性中性粒細胞減少症	VPS45
Pontocerebellar Hypoplasia, Type 2E	2E 型橋腦小腦發育不全	VPS53
Pontocerebellar Hypoplasia, Type 1A	1A 型腦橋小腦發育不全	VRK1
Microphthalmia/Anophthalmia, VSX2-Related	小眼畸形/無眼畸形 (VSX2 相關)	VSX2
Von Willebrand disease	血管性血友病	VWF
Wiskott-Aldrich syndrome, X-Linked	X 染色體Wiskott-Aldrich 綜合症	WAS
Progressive Pseudorheumatoid Dysplasia	進行性假性類風濕發育不良	WISP3
Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge S	Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge	WNT10A
Werner Syndrome	Werner綜合症	WRN
Xeroderma pigmentosum Group A	著色性乾皮病A組	XPA
Xeroderma Pigmentosum Group C	著色性乾皮病C組	XPC
Spastic Paraplegia Type 15	15 型痙攣性截癱	ZFYVE26

Last Updated: Jan 2023